Case Report

Oculoauriculovertebral Spectrum with Radial Anomaly in Child

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Abstract

Oculoauriculovertebral spectrum (OAVS) or Goldenhar syndrome is a wide spectrum of congenital anomalies that involves structures arising from the first and second branchial arches. It is characterized by a wide spectrum of symptoms and physical features. These abnormalities mainly involve the cheekbones, jaws, mouth, ears, eyes, or vertebrae. Other conditions with ear and/or radial involvement, such as, the Nager syndrome, Holt–Oram syndrome, Radial–renal syndrome, facioauriculoradial dysplasia, Fanconi anemia, and Vertebral, Anal atresia, Cardiac, Trachea, Esophageal, Renal, and Limb (VACTERL) association should be considered for differential diagnosis. Here we report a child who had facial asymmetry, microsomia, microtia, congenital facial nerve palsy, conductive hearing loss, skin tags, iris coloboma, and preaxial polydactyly.

Keywords: Goldenhar syndrome, hearing loss, hemifacial microsomia, radial defect

Introduction

In 1952, Goldenhar described a syndrome that presents with a combination of several anomalies, such as, dermal epibulbar tumors, periauricular appendices, and malformation of the ears. Later on, this condition also presented with heart diseases and hypoplasia of the zygomatic, mandibular, and maxillary bones. Oculoauriculovertebral spectrum (OAVS) presents with an estimated prevalence that ranges from 1 to 5,600 in 45,000 of live-births, and is considered the result of a blastogenesis defect that particularly involves the structures originating from the first branchial arches.[1,2] In approximately 5 to 15% of the cases, mild mental retardation may also be present. The anomalies observed in OAVS are hypoplasia of the thumb and hypoplasia and/or agenesis of the radio.[3,4] To date, about 32 cases of OAVS and radial defects have been reported.[5] Here we report a child who had OVAS with iris coloboma and a radial defect.

Case Report

An eight-year-old male child presented to the Pediatric Outpatient Department with complaints of facial asymmetry and polydactyly with bilateral microtia since birth. The child was born by vaginal delivery at a government hospital, with a birth weight of 2.5 kg. He was the second born. There was no family history of malformations. No mental retardation was detected during examination. The child showed undernourishment and weighed 15 kg (<fiftieth percentile) and a height of 110 cm (<fiftieth percentile). On physical examination, facial asymmetry, right side infranuclear facial palsy, bilateral microtia with bilateral ear tag [Figure 1], short neck, high arched palate, and skin tags on the nape of the neck were present. Dental caries were present, with normal dental development. There was lower motor neuron weakness of the facial nerve on the right side, manifested by an inability to close the right eye and loss of a nasolabial fold on the same side. Asymmetric crying facies was noted with the right lower lip being pulled down while crying or chewing. Left eye closure and nasolabial fold were normal. Left preaxial polydactyly was present [Figure 2], whereas, the other hand was normal. The length of both the hands was equal. His parents were normal on clinical examination. The first and third of the children, both boys, were normal.

On ophthalmological examination, the only positive finding was presence of a right-sided iris coloboma. Cardiac auscultation revealed that S1 and S2 were normal and a soft systolic murmur was present in the pulmonary area. No other abnormality

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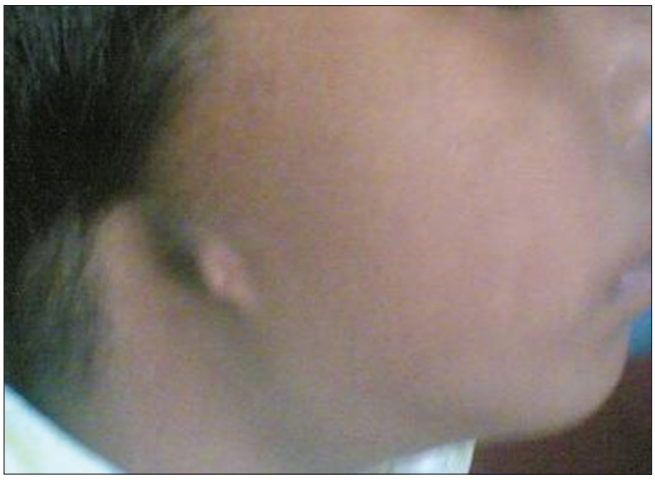

Figure 1: Microtia with preauricular tag

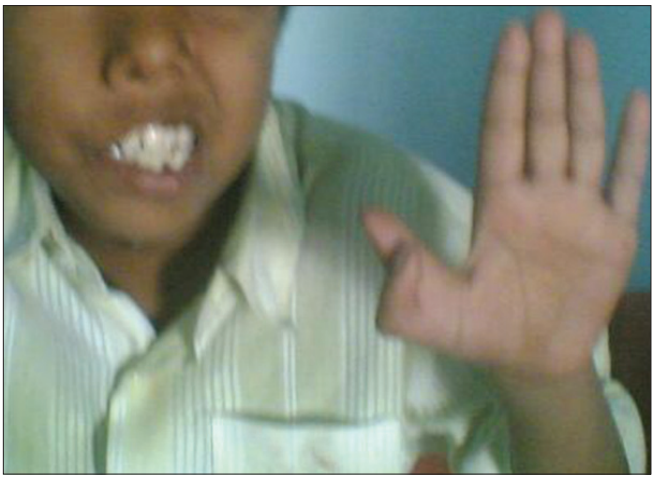

Figure 2: Facial asymmetry with left preaxial polydactyly

was detected on systemic examination. The patient showed moderate-to-severe bilateral conductive hearing loss on audiometry. Abdominal sonography was normal. Renal function tests, liver function tests, and thyroid function tests were normal. Echocardiography and Color Doppler did not detect any cardiac lesion. X-ray examination of the skull and vertebral column did not show any vertebral abnormality. A computed tomography (CT) scan of the temporal bone revealed bony atresia of the external auditory canal, with absence of mastoid pneumatization of the right ear and left ear, showing auricular tags with bony atresia of the external auditory canal, with a deformed ossicular chain.

Discussion

OAVS or Goldenhar syndrome is characterized by a variable degree of uni-or bilateral involvement of the craniofacial structures involving the first and second branchial arches, ocular anomalies, and a vertebral defect. The OAVS, with radial defects, characterizes a subset within the OAVS, mainly involving the first uni-or bilateral branchial arches and limb primordium.[5] The main signs include ear malformations, facial asymmetry, mandible hypoplasia, and a radial defect.

The etiology is not known, but various theories suggest that exposure to viruses during pregnancy, abnormal vascular supply to the first arch, and abnormality of mesoblastic development are reponsible. Some author reported that the disorder may be due to multifactorial inheritance.[6]

Ear anomalies, including preauricular tags, microtia anotia, and aural fistulae are reported. Wang, *et al.*[7] reported a 46% incidence of conductive hearing loss in their study. In our case, bilateral microtia with bilateral preauricular tags were observed, with an accompanying bilateral conductive hearing loss.

Craniofacial abnormalities may include malar hypoplasia, maxillary and mandibular hypoplasia, and temporal hypoplasia, incomplete development of the muscles of the face, macrostomia, cleft palate, cleft lip, and abnormalities of the teeth. Wang *et al.*[7] reported the incidence of facial nerve palsy in patients with craniofacial abnormalities to be 33.3%. In most cases, the etiological investigations of facial nerve palsy revealed an absence of physical or radiographic findings. The MRI of the temporal bone revealed normal components of the facial nerve and facial canal in our case. Ocular abnormalities included epibulbar dermoids or lipodermoids, unilateral microphthalmia, and upper eyelid coloboma. In our case right side iris coloboma was present.

Vendramini, *et al.*[5] reported that the most common anomalies observed in OAVS were hypoplasia of the thumb and hypoplasia and/or agenesis of the radio. The radial defects observed with less frequency were thumb agenesis, preaxial polydactyly, and triphalangeal thumb.[5] In our case left-side preaxial polydactyly was observed.

Congenital heart diseases are seen in 40 to 60% of the patients, but no cardiovascular abnormalities was found in our case. Renal problems are usually asymmetric and unilateral in 70% of the cases. The renal malformation like renal agenesis, ectopic kidney, urethral duplication, and vesicoureteric reflex was ruled out by ultrasonography, in our case. Vertebral anomalies like hemivertebrae and hypoplasia of the cervical vertebrae and cervical fusion were absent in this case.[4-7]

Our patient had facial asymmetry, microsomia, microtia, congenital facial nerve palsy, skin tags iris coloboma, and preaxial polydactyly. Surgical repair was often recommended for facial tags and macrostomia within the first year. Good oral hygiene was especially important for children with OAVS. Children should have good dental care. Orthodontic evaluations were important, to assess for missing teeth, dental crowding, jaw growth, and dental occlusion. All infants with OAVS should have a diagnostic hearing evaluation brainstem auditory evoked response (BAER) within the first six months of life. The structural anomalies of the eyes and ears could be corrected by plastic surgery. Orthodontic treatment could be required for malocclusion teeth. The use of hearing aids is needed early in life, in case of conductive deafness. In a

patient with underdevelopment of the lower jaw, reconstruction could be done with rib bone graft, and an underdeveloped mandible could be is lengthened by a bone distraction device. Reconstruction of the external ear could be performed at the age of six to eight years, whereas, the reconstructed asymmetry for cheek fullness could be done at eight to ten years of age. The surgical procedure involved temporarily wiring the jaws together during the operation.[8-9] In conclusion, in very rare cases OVAS is associated with a radial defect.

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